

Birth Defects Prevalence and Mortality

Q: What are the trends in human disease and conditions for which environmental contaminants may be a risk factors including across population subgroups and geographic regions?

The above question pertains to all 'Human Disease and Conditions ' Indicators, however, the information on these pages (overview, graphics, references and metadata) relates specifically to "Birth Defects Prevalence and Mortality". Use the right side drop list to view the other related indicators on this question.

Introduction

Birth defects are structural or functional anomalies causing physical or mental disability, some of which can be fatal. Although birth defects are the leading cause of infant mortality (deaths occurring to those under 1 year of age) in the U.S., the cause is unknown for approximately 70 percent of all cases (Infant Mortality indicator) (CDC, 2005). Many different factors may be associated with the development of birth defects, such as genetic and/or chromosomal aberrations, *in utero* exposure to viruses or bacteria, uncontrolled maternal diabetes, maternal cigarette smoke, maternal use of drugs and alcohol during pregnancy, and prenatal exposure to chemicals. All of these factors may influence normal infant growth or development, resulting in different types of birth defects (NICHD, 2006).

This indicator presents birth defects prevalence at birth for five specified congenital anomalies (anencephaly, cleft lip or palate, Down syndrome, omphalocele or gastroschisis, and spina bifida or meningomyelocele) that are consistently reported on both the 2003 (revised) and 1989 U.S. Standard Certificates of Live Birth and mortality rates among infants in the U.S. as recorded in the National Vital Statistics System (NVSS), which registers virtually all births and deaths nationwide. Data collection began in 1933 and is available through 2008. Birth defects data are collected on death certificates from all 50 states and the District of Columbia and recorded on birth certificates for 49 states and the District of Columbia. Reported race and ethnicity data are based on the race and ethnicity of the mother.

What The Data Show

Exhibit 5-36 presents the prevalence of live births with identified specific congenital anomalies (i.e., birth defects) between 1999 and 2008. The rates of cleft lip/palate were the highest of the five reported anomalies followed by Down syndrome, omphalocele or gastroschisis, spina bifida or meningomyelocele, and anencephaly. Rates for cleft lip or palate and spina bifida or meningomyelocele decreased slightly over the last decade. Rates for the other three fluctuated over time with a slight increase in rates since 1999.

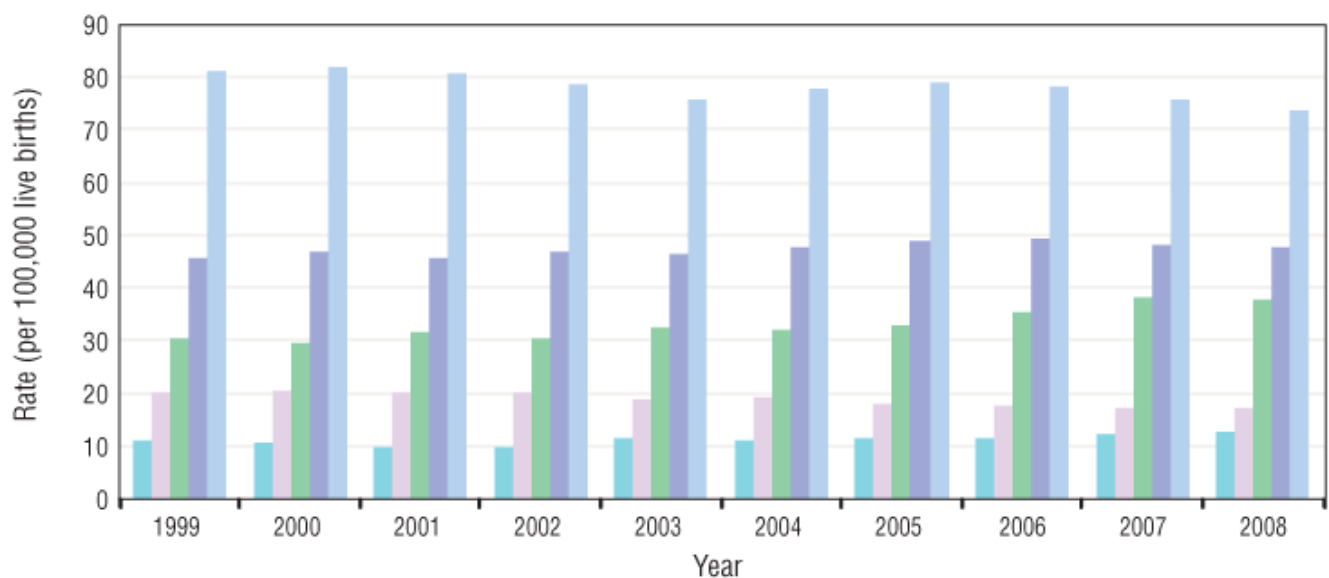
Rates for certain types of anomalies differ widely with maternal age. For example, in 2008 as in past years, infants of the youngest mothers (under 20 years of age) have the highest rates for omphalocele or gastroschisis, a defect or abnormality of the anterior abdominal wall (100.6 per 100,000 live births); infants of mothers age 40-54 years have the highest rates for Down syndrome (369.6 per 100,000 live births). (Data not shown.)

Birth defects continue to be the leading cause of infant mortality, accounting for 5,785 (19.9 percent) of the 29,138 infant deaths in 2007 (Exhibit 5-16, Infant Mortality indicator). Between 1979

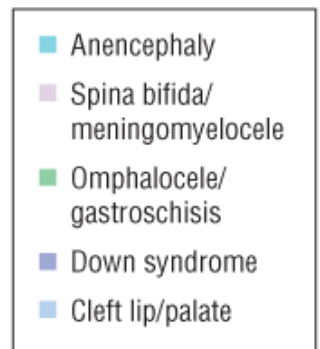
and 1998, a decline in the national birth defects mortality rate has been observed, ranging from 255.4 per 100,000 live births in 1979 to 157.6 per 100,000 live births in 1998. From 1999 to 2007, the birth defects mortality rates ranged from a high of 141.5 in 2000 to a low of 134.0 per 100,000 live births in 2007. (Data not shown.)

Asian or Pacific Islanders consistently had the lowest rates of mortality from birth defects among the reported racial groups with male and female Asian or Pacific Islander infants having mortality rates from birth defects of 87.6 and 81.2 per 100,000, respectively in 2007. Birth defects mortality was consistently higher among black infants than white infants. In 2007, for example, mortality attributed to birth defects among black male and female infants was 169.5 and 167.0 per 100,000 infants, respectively; among white male and female infants, it was 136.4 and 124.1 per 100,000 infants, respectively. (Data not shown.)

Exhibit 5-36. Rate of live births in the U.S. with specific birth defects (congenital anomalies), 1999-2008



Data source: NCHS, 2001; 2002a,b; 2003; 2005; 2006; 2007; 2009; 2010a,b



Limitations

- In order to enable comparisons over time, this indicator represents only a subset of possible birth defects. This is necessary because of changes made to the U.S. Standard Certificate of Live Birth in 2003. Consistent with NCHS reporting in its recent “Births: Final Data” publications, only the five congenital anomalies reported on both the 2003 and 1989 U.S. Standard Certificates of Live Birth are included (i.e., anencephaly, cleft lip or palate, Down syndrome, omphalocele or gastroschisis, and spina bifida or meningomyelocele).

- Because some birth defects are not recognized immediately, they are often underreported on both birth and death certificates (Friis and Sellers, 1999). Many anomalies are hard to detect at birth, which limits early ascertainment and complete reporting. The most serious and/or apparent anomalies are more likely to be identified and reported prior to hospital discharge (Honein et al., 2001). Research shows that the NVSS birth records can produce prevalence estimates that are 2 to 3 times lower than those based on ascertainment of congenital defects using records from active surveillance efforts (Parker et al., 2010). The congenital anomalies reported on birth certificates are rare events. Since a small change in the number of anomalies reported can result in a relatively large change in rates, caution should also be used in comparing yearly rates for a specific anomaly.
- Birth defects mortality rates are based on underlying cause of death as entered on a death certificate by a physician. Incorrect coding and low rates of autopsies that confirm the cause of death may occur. Additionally, some individuals may have had competing causes of death. “When more than one cause or condition is entered by the physician, the underlying cause is determined by the sequence of conditions on the certificate, provisions of the ICD [International Classification of Diseases], and associated selection rules and modifications” (CDC, n.d.). Consequently, some misclassification of reported mortality might occur in individuals with competing causes of death, as well as underreporting of some birth defects as the cause of death.
- The International Classification of Diseases 9th Revision (ICD-9) codes were used to specify underlying cause of death for years 1979-1998. Beginning in 1999, cause of death is specified with the International Classification of Diseases 10th Revision (ICD-10) codes. The two revisions differ substantially, and to prevent confusion about the significance of any specific disease code, data queries are separate. The relatively large difference between birth defects mortality rates reported from 1979 through 1998 and those reported beginning in 1999 may be due to some changes in the criteria used to report birth defects mortality during the switch from ICD-9 to ICD-10.

Data Sources

The birth defects rate data used for this indicator are from National Vital Statistics Reports published by the Centers for Disease Control and Prevention's National Center for Health Statistics (NCHS, 2001, 2002a,b, 2003, 2005, 2006, 2007, 2009, 2010a,b). The birth defects mortality data were obtained from a published report by the National Center for Health Statistics (NCHS, 2010c) and from CDC's compressed mortality files (underlying cause of death), accessed via CDC WONDER (CDC, 2011), at <http://wonder.cdc.gov>.

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NCHS. 2006. Births: Final data for 2004. National Vital Statistics Reports 55(1). http://www.cdc.gov/nchs/data/nvsr/nvsr55/nvsr55_01.pdf See Table 25.

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NCHS. 2003. Births: Final data for 2002. National Vital Statistics Reports 52(10). http://www.cdc.gov/nchs/data/nvsr/nvsr52/nvsr52_10.pdf See Table 49.

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NCHS. 2002b. Births: Final data for 2000. National Vital Statistics Reports 50(5). http://www.cdc.gov/nchs/data/nvsr/nvsr50/nvsr50_05.pdf See Table 49.

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NICHHD (National Institute of Child Health and Human Development). 2006. Birth defects and developmental disabilities. Accessed September 2007. <http://www.nichd.nih.gov/womenshealth/research/pregbirth/birthdefects.cfm>

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